MEFV gene mutation analysis in patient with PFAPA

Abstract :
Introduction : syndrome PFAPA (periodic fever, aphthous mouth, pharyngitis and adenitis) is a clinical syndrome that usually occur in children younger than 5 years. This disease is a sporadic disease, and periodic attacks of inflammation can be seen. Specific gen mutation has not seen in PFAPA. However,there is the possibility that involved genes in other autoinflammatory diseases play a role in it's pathogenesis. Thus, according to the clinical similarities between PFAPA and FMF, MEFV gene may be involved in PFAPA. The purpose of this study is to evaluate the MEFV gene mutations in patients with PFAPA syndrome .
Materials and Methods: This is a descriptive-analytical-sectional study and was performed on all patients with PFAPA syndrome that referred to rheumatology clinic of Imam Khomeini Hospital in Ardabil is 1391. Questions are asked from patients about demographic information, symptoms, age of onset, duration of symptoms, and ... . The patient are referred to a genetics laboratory and genetic tests are avaluated. The results of the genetic test with information of check lists are imported in Statistical analysis programs. The results are analyzed with SPSS v16.
Results: In this study, 21 patients with PFAPA were entered into the study, that 15 patients (71.4%) were male and 6 patients (28.6%) were female, and the mean age of patients was 8 ± 3.17 years. The majority of patients are living in Ardebil with 95.2%. The most common complaint were Fever with 95.2%, and the mean age of onset of symptoms in these children were 2.72 ± 1.52 years, respectively. The mean duration of fever in children were 3.57 ± 1.64 days, the mean duration of the disease were 4.23 ± 1.56 days and the mean duration of recovery from disease in these children were 32.52 ± 20.53 days. In a systematic review of the children's symptoms were observed fever with 100%, chills with 61.90%, weakness and lethargy with 71.42%, headache in 6 patients (28.6%), aphthous in 14 patients (66.6%), Pharyngeal erythema in 5 children (23.8%), red-eye in 1 patient (4.8%), Arthritis in 1 Child (4.8%), cough in 5 patients (23.8%), dyspnea in 2 patients (9.5%), palpitations with 9.5%, abdominal pain in 12 cases (57.14%) and myalgia in 13 patients (61.9%). This stody found a familial history of periodic fever in 3 patient [1)periodic fever in patient's brother, 2)FMF in patient's father, 3) FMF in patient's mother]. Among the 21 children with the PFAPA syndrome were 8 children (38.09%) with common MEFV gene mutations. In 7 child was Compound Heterozygotes and in 1 child was homozygotes. Also in review of alleles were observed the 4 allele (44.4%) M694V, 3 allele (33.3%) V726A, 1 allele (11.15%) E148Q and 1 allele K694R (11.15 percen%t).
Conclusions: considering that 38 percent of patients had common MEFV gene mutations, it seems that genetic basis of FMF be involved in PFAPA. Also in this stody, calculated gaslini diagnostic score showed this scoring to predict the probability of MEFV gene mutation (12 common mutation) in these patients is not worthwhile.
Keywords : Syndrome PFAPA, MEFV gene mutation.